

AMENDMENTS TO THE SPECIFICATION

On page 3, please replace the paragraph spanning lines 6-10 with the following paragraph:

Thus, in a first embodiment, the invention provides a nucleic acid (SEQ ~~ID~~ NO: 1 and 2) encoding MCOLN1 (SEQ ID NO:3); the MCOLN1 protein is also called "mucolipin". In particular, the invention provides an isolated *MCOLN1* gene (this gene had previously been referred to as MCL4) including non-coding and non-transcribed sequences. The invention also provides a *MCOLN1* cDNA, *i.e.*, free of introns.

On page 6, please replace the paragraph spanning lines 10-19 with the following paragraph:

The present invention is based, in part, on additional 9 AJ and 5 NJ families that were studied in addition to the studies reported in Slaugenhaupt et al. (Am. J. Human Genet. 1999, 65:777-8). We conducted a detailed haplotype analysis in order to pinpoint the gene location and determine the probable number of mutations. ~~There are 5 unique haplotypes in the AJ population, the major and minor haplotypes are present on 73% and 23% of chromosomes, respectively.~~
There are 5 unique haplotypes in the AJ population. The major and minor haplotypes are present on 73% and 23% of chromosomes, respectively. The remaining three haplotypes were only seen once; in two cases coupled with the major and once with the minor haplotype. Analysis of the 5 NJ families yielded an additional 7 unique haplotypes, suggesting that there may be as many as 12 independent mutations. Linkage disequilibrium analysis of the 2 common haplotypes enabled us to narrow the candidate region to 143 kb and we constructed a detailed transcript map of this interval.

On pages 6-7, please replace the paragraph spanning lines 26-29 on page 6 and continuing through lines 1-2 on page 7 with the following paragraph:

The term "functional MCOLN1" refers to an MCOLN1 that functions in a cell, *e.g.*, plays a role as a TRP channel or a receptor-stimulated cation channel. Evidence of MCOLN1 function can be detected by various methods. MCOLN1 functions include, but are not limited to, HCl secretion, ion channel activity, and secretion of solutes from ~~intreacellular~~ intracellular vesicles. Other MCOLN1 functions include, but are not limited to, binding with MCOLN1-specific antibodies.

On page 7 of the application, please replace the paragraph spanning lines 9-14 with the following paragraph:

MLIV is a disease with phenotypic characteristics similar to mucopolysaccharidosis without sugar in the urine, with features that include growth and mental retardation, corneal clouding and ~~typesonal~~ lysosomal inclusions. MLIV is in the category of channelopathies ~~channelopathies~~, *i.e.*, ion channel defects. Thus, the present invention concerns mucopolipidosis and certain other conditions that result from an ion channel defect, wherein the ion channel is MCOLN1.

On page 13, please replace the paragraph spanning lines 1-4 with the following paragraph:

A coding sequence is "under the control of" or "operatively associated with" of transcriptional and translational control sequences in a cell when RNA polymerase transcribes the coding sequence into mRNA, which is then trans-RNA spliced (if it contains introns) and translated into the protein encoded by the coding sequence.

In the heading preceding [0079] of the published application, we note an error that is not present in our submission. In accordance please replace this heading with the following heading:

Expression of MCOLN1 ~~Polypeptides~~ Polypeptides

In Table 1, presented on pages 21-22, and again on page 42, of the specification, please replace the second entry in the last column of row 8 {corresponding to Haplotype 42(NJ)} as follows:

D362T D362Y

In paragraph [0068] of the published application, but not in the corresponding paragraph of the application, some of the text is erroneously represented in bold font. Please change this text back to plain font.

Applicants note that they have not added new matter by way of these amendments.